

ABSTRACT

A novel gene defining a novel human UDP-GlcNAc: Gal/GlcNAc β 1-3GalNAc α 1, 6GlcNAc-transferase, termed C2/4GnT, with unique enzymatic properties is disclosed. The enzymatic activity of C2/4GnT is shown to be distinct from that of previously identified enzymes of this gene family. The invention discloses isolated DNA molecules and DNA constructs encoding C2/4GnT and derivatives thereof by way of amino acid deletion, substitution or insertion exhibiting C2/4GnT activity, as well as cloning and expression vectors including such DNA, cells transfected with the vectors, and recombinant methods for providing C2/4GnT. The enzyme C2/4GnT and C2/4GnT-active derivatives thereof are disclosed, in particular soluble derivatives comprising the catalytically active domain of C2/4GnT. Further, the invention discloses methods of obtaining 1,6-N- acetyl glucosaminyl glycosylated saccharides, glycopeptides or glycoproteins by use of an enzymically active C2/4GnT protein or fusion protein thereof or by using cells stably transfected with a vector including DNA encoding an enzymatically active C2/4GnT protein as an expression system for recombinant production of such glycopeptides or glycoproteins. Also a method for the identification for the identification of DNA sequence variations in the C2/4GnT gene by isolating DNA from a patient, amplifying C2/4GnT-coding exons by PCR, and detecting the presence of DNA sequence variation are disclosed.